Copy number variation (CNV) refers to a large range of DNA rearrangement (deletion or duplication) which cannot be detected by conventional sequencing analysis. If sequencing test is negative, CNV analysis is highly recommended otherwise the testing result is inconclusive. Array-based comparative genomic hybridization (aCGH) is one of the best tools available to analyze CNV.

The John Welsh Cardiovascular Diagnostic Laboratory offers a CNV test that is specifically designed to enrich 500 genes related to cardiovascular diseases using Agilent aCGH platform. This test has been validated in our laboratory on many samples with known clinically relevant CNVs. It is designed to detect losses and gains representing deletions, duplications or amplifications of the cardiovascular genes included. This assay will not detect balanced translocations, inversions, or point mutations that may be responsible for the clinical phenotype.

**REASONS FOR REFERRAL**

Molecular confirmation of the diagnosis of certain cardiovascular diseases.

**METHODOLOGY**

Genomic DNA is analyzed for CNVs by aCGH (Agilent 4x180k).

**SERVICE FEES**

<table>
<thead>
<tr>
<th></th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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</thead>
<tbody>
<tr>
<td>Single Gene</td>
<td>$1,200 per sample</td>
<td>81228</td>
</tr>
<tr>
<td>Multiple Genes or Panel (s)</td>
<td>$1,500 per sample;</td>
<td>81228</td>
</tr>
</tbody>
</table>

**SENSITIVITY**

This target enriched array can detect as small as 1 kb deletion or duplication in exonic regions.

**SPECIMEN REQUIREMENTS**

**Blood (preferred):** EDTA (purple-top) tubes: *Adult*: 5 cc  *Child*: 5 cc  *Infant*: 2-3 cc  
**Tissue:** Frozen (preferred), RNA*later*,  
**Other Body Fluids:** Call to inquire